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PKHD1 wt Allele

National Cancer Institute

Source

National Cancer Institute. *PKHD1 wt Allele*. NCI Thesaurus. Code C150299.

Human PKHD1 wild-type allele is located within 6p12.3-p12.2 and is approximately 473 kb in length. This allele, which encodes fibrocystin protein, may play a role in kidney and biliary development and in receptor signaling. Mutations in this gene are associated with autosomal recessive polycystic kidney disease (polycystic kidney and hepatic disease 1; polycystic kidney disease 4).